



MURCS association and anorectal malformation: Case report of a female newborn



Sofia Morão ^{a,*}, Fernando Chaves ^b, Daniel Virella ^b, Fátima Alves ^c, Rui Alves ^a, João Pascoal ^a

^a Pediatric Surgery Department, Hospital Dona Estefânia, Centro Hospitalar Lisboa Central, Portugal

^b Neonatal Intensive Care Unit, Hospital Dona Estefânia, Centro Hospitalar Lisboa Central, Portugal

^c Pediatric Surgery Department, Urology Unit, Hospital Dona Estefânia, Centro Hospitalar Lisboa Central, Portugal

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ABSTRACT

MURCS association is rare, first described by Duncan et al. in 1979, including Müllerian duct aplasia, renal aplasia and cervicothoracic somite dysplasia. Levitt and Peña described in 2007 a classification of syndromic anorectal malformation (ARM) that associates these two entities. The reported case is the first one described in neonatal period. We describe a case of a female newborn with suspected diagnosis of anorectal and renal malformations. Physical and radiologic investigation revealed agenesis of sacrum and coccyx, tethered cord, left multicystic renal dysplasia, absence of vaginal orifice and hymen, normally placed urethral orifice and abnormal anal opening at the vaginal introitus as a rectovestibular type fistula. Also, she had right uterine, tube and ovary agenesis with a normal 46, XX female karyotype. A left diversing colostomy was done in first day of life and four months later, was performed a posterior sagittal anorectoplasty (PSARP), with intra-operative identification of a duplication of the distal rectum (related with caudal regression syndrome type 2). There were no complications in postoperative period. A staged management strategy is a viable option avoiding further complications in an already poor prognosis situation.

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1. Introduction

MURCS association, first described by Duncan et al., in 1979, is a rare and nonrandom constellation of findings that includes Müllerian duct aplasia, renal aplasia and cervicothoracic somite dysplasia [1]. Anorectal malformations (ARM) comprise a wide spectrum of diseases that involve the distal anus and rectum as well as the urinary and genital tracts [2,3]. Levitt and Peña described in 2007 a classification of syndromic ARM that associates these two entities [3]. As it is extremely rare, high suspicion is essential for diagnosis and surgical orientation. We present a case diagnosed in neonatal period which poses several concerns regarding growth, urinary and bowel control.

2. Case report

A 40-year-old G3P3 presented at 39 weeks and 3 days gestation with premature rupture of membranes 36 h before delivery with clear amniotic fluid. Fetal ultrasound at 34 weeks of gestation suggested presence of multicyst left kidney. Because of suspected fetal distress, a caesarian delivery was performed. After delivery, the neonate needed non-invasive ventilation with quick recovery of heart rate. Apgar scores were 6 at 1 min but 8 at 5 min. The neonate had female phenotypic appearance and weighted 2580 g. Physical examination at that point revealed absence of anal orifice so it was decided to transfer her to a tertiary care pediatric hospital. On presentation, the neonate was hemodynamically stable. The lateral abdominal x-ray revealed agenesis of sacrum and coccyx (Fig. 1).

Renal and pelvic ultrasound suggested presence of tethered cord and showed left multicystic renal dysplasia (Fig. 2).

Transfontanelar ultrasound didn't reveal any associated malformations. The echocardiogram demonstrated patent ductus arteriosus, tricuspid and pulmonary regurgitation. After observation by a pediatric surgeon, who confirmed the presence of ARM

* Corresponding author. Pediatric Surgery Department, Hospital Dona Estefânia, Centro Hospitalar Lisboa Central, Rua Jacinta Marto, 1169-045, Lisboa, Portugal.

E-mail address: sophia.morao@gmail.com (S. Morão).

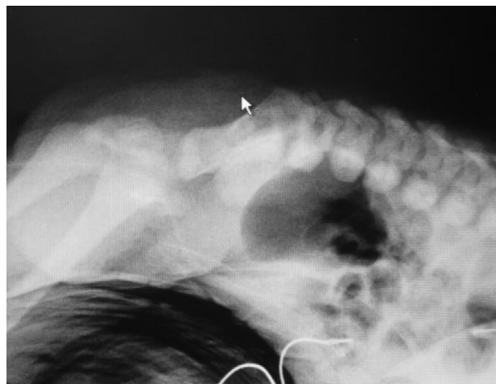


Fig. 1. Lateral x-ray.

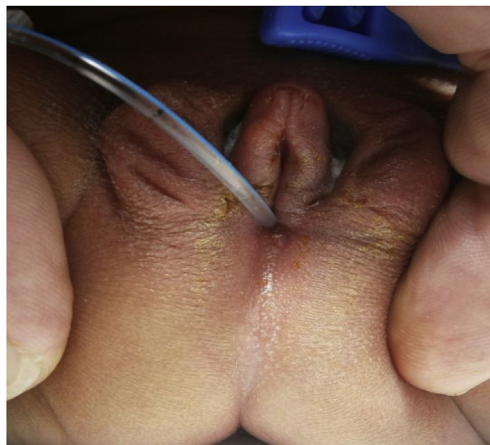


Fig. 3. Perineal inspection.

and it was decided to perform a left diverting colostomy. Under general anesthesia, the genital inspection, performed before the procedure, detected the absence of normal vaginal orifice and hymen, normally placed and permeable urethral orifice, and finally, an abnormal anal opening at the vaginal introitus as a variation of a rectovestibular type fistula (Figs. 3 and 4).

Intraoperatively, it was observed right uterine, tube and ovary agenesis with left structures intact (Figs. 5–7). Further investigation showed a normal 46, XX female karyotype, right grade I vesicoureteral reflux (VUR) in cystography (according to the International Reflux Committee classification) (Fig. 8), left kidney with 2% function in MAG3 renogram. The distal pressure colostogram revealed the presence of an abnormal reflux of contrast to the vaginal lumen (Fig. 9).

At four months of age, it was performed a PSARP procedure with interoperative identification of a 2-cm-long tubular duplication of rectum (Figs. 10–12). There were no complications in the post-operative period. At the most recent appointment, the child is one year old and is growing well; she is under anal dilatation scheme. Colostomy is working properly and she wears diapers (Fig. 13).

3. Discussion

In females, fallopian tubes, uterus and upper third of vagina

develop from Müllerian ducts while lower two thirds of vagina and vaginal vestibule develop from lower part of definitive urogenital sinus [4]. Malformations of the uterus or vagina are attributed to abnormalities of fusion or regression of the caudal ends of the müllerian ducts.

In this case, we have right ovary, fallopian tube and hemi-uterus agenesis which excludes Mayer-Rokitansky-Küster-Hauser syndrome (vaginal agenesis, uterine malformation but normal tubes and ovaries). Moreover, right müllerian structures agenesis with left renal dysplasia excludes Herlyn-Werner-Wunderlich syndrome (müllerian ducts malformations with ipsilateral renal anomaly).

MURCS association is named when nonrandom combination of malformations (müllerian, renal and cervicothoracic) is formed together. The etiology of MURCS association is not known. The karyotype was normal in all investigated cases, including ours [1,5]. The presented case has all the classical findings described by Duncan et al. in 1979, except for vertebral anomalies which are seen in lower part of column. However, as it is mentioned on literature, identification of one component of the MURCS association suggests the presence of the other associated anomalies which may not be noticed until later in life. The child described in this paper has only 13 months of age. Duncan hypothesized about a close spatial relationship at the end of the fourth week of fetal life between cervical-upper thoracic somites, arm buds, and pronephric ducts. An error at this stage would cause the maldevelopment of vertebrae and kidneys and organ systems originating from müllerian ducts [1].

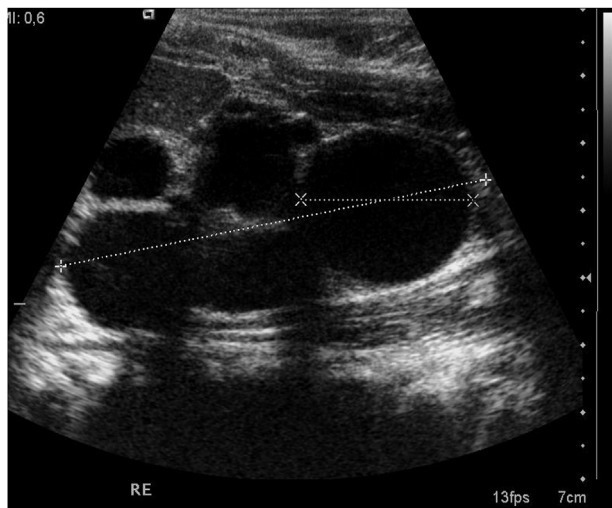


Fig. 2. Renal ultrasound.

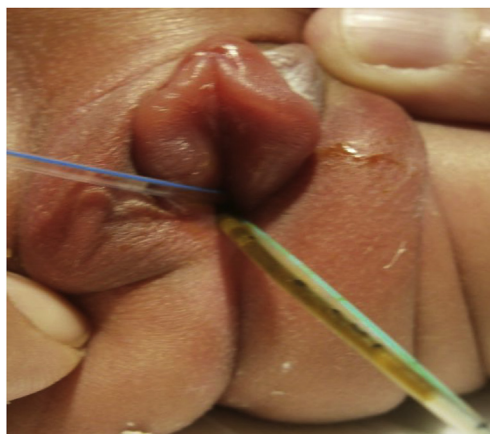


Fig. 4. Perineal inspection.

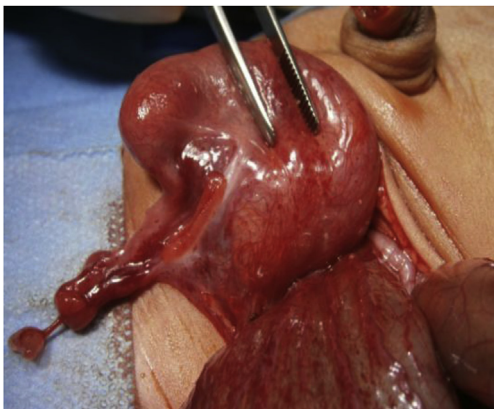


Fig. 5. Left ovary, tube and hemiuterus. Descending diversing colostomy.



Fig. 6. Left ovary, tube and hemiuterus. Descending diversing colostomy.

ARM comprise a wide spectrum of diseases that involve the distal anus and rectum as well as the urinary and genital tracts. They occur in approximately 1 in 5000 live births. Defects range from the very minor and easily treated with an excellent functional prognosis to those that are complex, difficult to manage, are often associated with other anomalies and have a poorer functional prognosis. ARM with rectovestibular type fistula is an intermediary type anomaly; it is the most common type in girls [2,3,5]. The

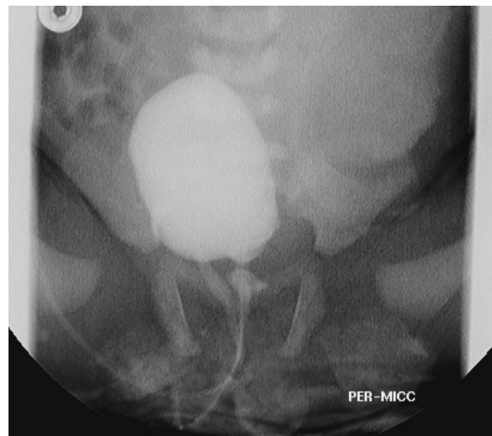


Fig. 8. Cystography.



Fig. 9. Colostogram.

genitourinary tract is the most serious and frequent site of associated defects in ARM (frequency varies from 20 to 54% depending on the source of reference) [2] but a combination with MURCS association is very rare. In 2007, Alberto Peña and Mark Levitt published a paper where they tried to group together defects that have



Fig. 7. Left ovary, tube and hemiuterus. Descending diversing colostomy.



Fig. 10. Posterior sagittal anorectoplasty. Presence of an endoluminal septum – tubular rectal duplication.

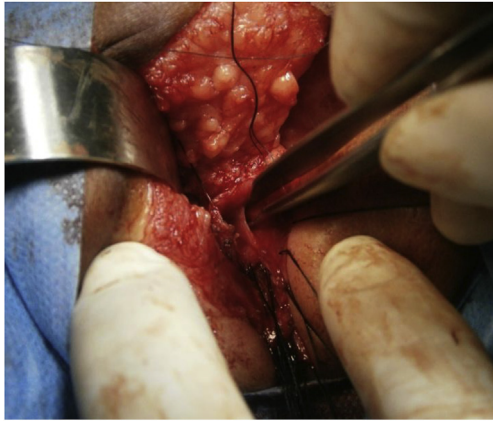


Fig. 11. Posterior sagittal anorectoplasty. Presence of an endoluminal septum – tubular rectal duplication.

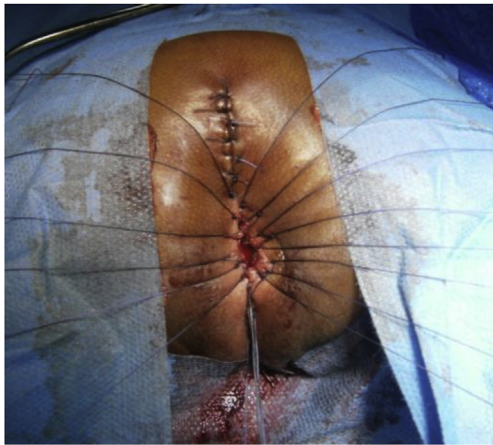


Fig. 12. Posterior sagittal anorectoplasty. Presence of an endoluminal septum – tubular rectal duplication.

common diagnostic, therapeutic and prognostic features. There, they describe a syndromic ARM that associates both ARM and MURCS association. Our patient had diagnosis of ARM with rectovestibular type fistula after observation by pediatric surgeon, but MURCS association was only considered after abdominal exploration during colostomy surgery. As far as we have seen in literature, this is a unique case diagnosed in neonatal period [5–10].

Spinal anomalies such as abnormal sacrum, presence of hemi-vertebra and tethered cord are frequent and associated with the worst functional prognosis. In our case, besides sacrum and coccyx agenesis and tethered cord, there was also found duplication of rectum, which has been associated with caudal regression syndrome. So, motor and sensory disturbances of the lower extremities are expected as well as fecal and urinary incontinence. Very likely, later in life, she will benefit from release of spinal cord by a neurosurgeon; she will need a bowel management program after closure of colostomy and proper management of neurogenic bladder [11,12]. Since early 80's, it is well established that, in this type of ARM, a decompressive and protective colostomy is performed in the neonatal period [2]. PSARP is executed some weeks later.

In this case, the child also has distal (partial) vaginal agenesis. We believe that there is no need to use distal rectum or sigmoid to create a neovagina as she has upper portion of vagina and using rectum as neovagina would disturb even more bowel control. At



Fig. 13. Perineum at one year of age.

our department, most of the patients that need vaginal reconstruction are divided in two groups: one group includes feminizing genitoplasty in children with congenital adrenal hyperplasia which is performed in early childhood or pre-scholar age; the other group includes cases of Wunderlich and Mayer-Rokitansky-Küster-Hauser syndromes as well as uterovaginal malformations as transverse or longitudinal vaginal septum, in which vaginoplasty is best performed in late infancy or at prepuberty. Our plan is performing mobilization of the native upper vagina associated with perineal skin flap (Fortunoff technique) [13] as a second procedure, accordingly with her somatic, neurologic and sexual development. Although it is described in literature that it is possible to perform correction of ARM and vaginal reconstruction at one time [14], avoiding reoperation in a fibrotic area, and despite being a tertiary referral hospital, our cumulated experience is to perform these procedures in separate times later in life.

Colostomy will be maintained until the desired size of anus is achieved.

4. Conclusion

It is obvious that prognosis of this child regarding bowel and urinary control is extremely poor. A staged management strategy was adopted with meticulous surgical repair trying to avoid complications and confer the best chance for a reasonable outcome.

References

- [1] Duncan PA, Shapiro LR, Stangel JJ, Klein RM, Addonizio JC. The MURCS association – müllerian duct aplasia, renal aplasia and cervicothoracic somite dysplasia. *J Pediatr* 1979, September;95(3):399–402.
- [2] Peña A. Atlas of surgical management of anorectal malformations. Springer-Verlag; 1990.
- [3] Levitt M, Peña A. Anorectal malformations – review. *Orphanet J Rare Dis* 2007;2:33.
- [4] Carlson BM. Human embryology and developmental biology – updated edition. third ed., vol. 16. Elsevier Mosby; 2004. p. 393–427.
- [5] Günşar C, Genç A, Şencan A, Dağlar Z, Alparslan O, Mir E. MURCS association and rectovestibular fistula: case report of a patient treated with one-stage posterior sagittal anorectoplasty and sigmoid loop vaginoplasty. *J Pediatr Surg* 2003;38(2):262–4.
- [6] Ramakrishna, Kotecha N, Patel CA, Pipavat R. MURCS Association: a rare association with patent ductus arteriosus and bicuspid aortic valve. *J Assoc Physicians India* 2013 November;61(11):832–4.
- [7] Braun-Quentin C, Billes C, Böwing B, Kotzot D. MURCS association: case report and review. *J Med Genet* 1996 July;33(7):618–20.

- [8] Akhter N, Begum BN, Newaz M. The MURCS association: mullerian duct aplasia, renal hypoplasia and cervicothoracic somite dysplasia – a case report. *Mymensingh Med J* 2015 July;24(3):600–5.
- [9] Saad Ganem A, López-Ortiz CG, Lejtik Alva CA, Quintero Bernal P, Zafra de la Rosa G, Gutiérrez Palomares V. MURCS association; case report. *Ginecol Obstet Mex* 2008 June;76(6):349–52.
- [10] Vergnes C, Cordier MP, Dubois R, Bouvier R, Cochat P. MURCS association: a challenging diagnosis. *Arch Pediatr* 2005 January;12(1):49–51.
- [11] Levitt M, Peña A. Fecal incontinence and constipation. In: *Ashcraft's pediatric surgery*. sixth ed., vol. 36. , Philadelphia: Saunders Elseviers; 2014. p. 515–24.
- [12] Cartwright P, Snow B, Wallis C. Bladder and urethra. In: *Ashcraft's pediatric surgery*. sixth ed., vol. 56. , Philadelphia: Saunders Elseviers; 2014. p. 749–61.
- [13] Fortunoff S, Lattimer JK, Edson M. Vaginoplasty technique for female pseudohermaphrodites. *Surg Gynecol Obstet* 1964;118:545–8.
- [14] Levitt M, Bischoff A, Breech L, Peña A. Rectovestibular fistula – rarely recognized associated gynecologic anomalies. *J Pediatr Surg* 2009;44:1261–7.